PLOD1 gene

procollagen-lysine,2-oxoglutarate 5-dioxygenase 1

Normal Function

The *PLOD1* gene provides instructions for making an enzyme called lysyl hydroxylase 1. This enzyme modifies an amino acid called lysine, which is one of the building blocks used to make proteins. Specifically, lysyl hydroxylase 1 converts lysine to a similar molecule, hydroxylysine, through a chemical reaction called hydroxylation. Hydroxylysine is commonly found in collagens, which are complex molecules that provide strength and support to many body tissues.

Hydroxylysine is essential for collagen molecules to form stable interactions, called cross-links, with one another in the spaces between cells. The cross-links result in the formation of very strong collagen fibers.

Health Conditions Related to Genetic Changes

Ehlers-Danlos syndrome

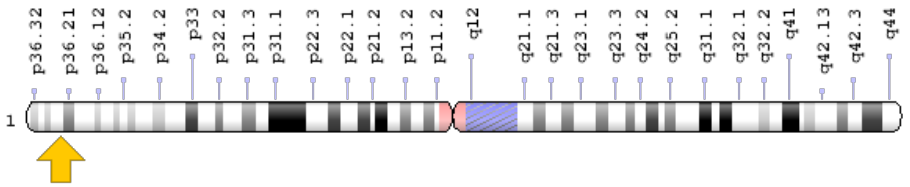
More than 30 mutations in the *PLOD1* gene have been found to cause a form of Ehlers-Danlos syndrome called the kyphoscoliosis type. Ehlers-Danlos syndrome is a group of disorders that affect the connective tissues that support the skin, bones, blood vessels, and many other organs and tissues. The kyphoscoliosis type is characterized by an unusually large range of joint movement (hypermobility) and severe, progressive curvature of the spine that can interfere with breathing.

The most common *PLOD1* gene mutation copies (duplicates) a large portion of the gene, resulting in the production of a nonfunctional version of the lysyl hydroxylase 1 enzyme. Several other mutations introduce premature stop signals that prevent the production of any functional enzyme. A loss of lysyl hydroxylase 1 activity greatly reduces the amount of hydroxylysine, which impairs cross-linking between collagen molecules. This disruption in the network of collagen fibers weakens connective tissues, causing the signs and symptoms of the kyphoscoliosis type of Ehlers-Danlos syndrome.

Chromosomal Location

Cytogenetic Location: 1p36.22, which is the short (p) arm of chromosome 1 at position 36.22

Molecular Location: base pairs 11,934,667 to 11,975,542 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- collagen lysyl hydroxylase
- LH
- LH1
- LLH
- lysine 2-oxoglutarate dioxygenase
- lysine hydroxylase
- lysyl hydroxylase
- PLOD
- PLOD1_HUMAN
- procollagen-L-lysine,2-oxoglutarate:oxygen oxidoreductase (5-hydroxylating)
- procollagen-lysine 1, 2-oxoglutarate 5-dioxygenase 1
- procollagen-lysine, 2-oxoglutarate 5-dioxygenase (lysine hydroxylase, Ehlers-Danlos syndrome type VI)
- procollagen-lysine, 2-oxoglutarate 5-dioxygenase 1
- protocollagen lysyl hydroxylase

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Collagen: The Fibrous Proteins of the Matrix
  https://www.ncbi.nlm.nih.gov/books/NBK26810/figure/A3558/

GeneReviews

- Ehlers-Danlos Syndrome, Kyphoscoliotic Form
  https://www.ncbi.nlm.nih.gov/books/NBK1462

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PLOD1%5BTIAB%5D%29+OR+%28lysyl+hydroxylase+1%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- PROCOLLAGEN-LYSINE, 2-OXOGLUTARATE 5-DIOXYGENASE
  http://omim.org/entry/153454

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_PLOD1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PLOD1%5Bgene%5D
- Ehlers-Danlos Syndrome Variant Database
  https://eds.gene.le.ac.uk/home.php?select_db=PLOD1
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9081
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5351
- UniProt
  http://www.uniprot.org/uniprot/Q02809

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